

Genome sequencing tests to help track rare diseases

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Patients with undiagnosed rare diseases suspected to be linked to genetic mutations can now undergo genome sequencing tests for free to find the cause of their illness.

The Hong Kong Genome Institute has so far recruited nine local patients and their families to join the scheme that was launched last year. This includes a 15-year-old girl suffering from kidney failure caused by an unknown reason, which is rare at such a young age.

Brian Chung Hon-Yin, chief scientific officer of HKGI, said

the girl, as well as eight other patients, have joined the scheme for free, hoping to find out the reason behind their illnesses by having their blood samples tested in Hong Kong.

Without genome sequencing technology, patients suffering from rare diseases need to consult an average of five doctors before a diagnosis, Chung said. They also have to wait for four years, spend HK\$74,500 and receive three misdiagnoses on average.

Chung stressed that the new

technology is crucial in identifying rare diseases, citing the example of his previous patient, 10-year-old Anna Chu, who is suffering from the rare Alstrom syndrome.

After noticing the girl's vision problems when she was only three to four months old, her parents took her to many eye specialists but still failed to identify the cause of her illness.

She was finally diagnosed with Alstrom syndrome after a genome sequencing test three years ago, at the age of seven.

“We had no idea about Anna's disease. We did not know what do to. It was like drifting in a dark ocean

with no direction. When we received the result, it was like land was finally in sight,” Anna's father said.

The data of patients who join the scheme will also be used to build a database of rare diseases and genetic mutations, Chung said.

“The current worldwide database on genetic disease mainly consists of the genes of white people in western countries,” Chung said. “We need to build our own database, which can help Hong Kong patients the most.”



Anna Chu, center left, and Brian Chung, inset.